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LABORATORY BULLETIN

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DEPARTMENT OF HEALTH & ENVIRONMENTAL SCIENCES, HELENA, MONTANA

No. 57 Editor David B. Lackman, Ph.D., Administrator, Laboratory Division
March 10, 1975

STATE DOCUMENTS

INFANT SCREENING TESTS FOR INBORN ERRORS OF METABOLISM - CONTINUED*
(PREVENTIVE MEDICINE AT WORK!)

On February 25, 1975, Dr. John Anderson, Director of the Montana State Department of Health and Environmental Sciences, signed the contract placing us in the OREGON METABOLIC DISEASE SCREENING PROGRAM. It is our hope that this will serve as a model for interstate cooperation. The concept of regionalization of services is becoming increasingly important for those of us who must operate from a relatively small population base. Montana's estimated population is 721,000 and, with respect to this program, we have about 11,500 live births a year. To make a comprehensive screening program like this practical, about 50,000 live births a year are required.

This is the final phase in the implementation of House Bill 261 which was signed by Governor Thomas L. Judge on March 8, 1973. It is now Section 69-6710 through 69-6713, Revised Codes of Montana 1947. Rules for implementation of the law were adopted by the Board of Health and Environmental Sciences on March 19, 1974. They are now Montana Administrative Codes pp 16-404 to 16-406, Sub-Chapter 6, Maternal and Child Health Bureau : 16-2.18 (6) -S1820 INFANT SCREENING TESTS.

Currently, tests mandated under provisions of the Codes are for phenylalanine, leucine (Maple Syrup Urine Disease), methionine, tyrosine, and galactose (galactosemia). At a meeting of an advisory committee on January 11, 1974, there were discussions of screening of newborn for hypothyroidism and mucopolysaccharidoses. Difficulties and uncertainties surrounding both of these areas dictated that they be held in abeyance. However, during my visit to Portland (it came off on January 21, 1975 as announced in Bulletin No. 56), I discovered that hypothyroid screening is just around the corner. Dr. Murphey, from Dr. Guthrie's department, who is spending a leave of absence in the Oregon laboratory, is working to develop a procedure which would be applicable to screening of infants for evidence of congenital hypothyroidism. Here is a recent quote from Dr. Guthrie : *"We certainly hope to include Larson's T_4 method for hypothyroidism, as the Quebec results (7 confirmed cases in 49,000 are claimed) plus 1/1800 in Toronto and one in an equal number in Pittsburgh all indicate that this screening procedure is indeed worthwhile."* I included a comment from the Massachusetts program in Bulletin No. 56 which also indicated that such screening would be worthwhile. Information from Dr. Pallister indicates that hypothyroidism in Montana is an important underlying cause for admissions to Boulder. Therefore, we are anticipating that the next test to be added in the program will be thyroid screening.

There is still no practical method of screening for mucopolysaccharidoses in a routine program. The Berry spot test used routinely causes confusion because it yields up to 30 percent false positives. This test is useful for checking the residual spot for mucopolysaccharides after thin-layer amino-acid chromatography. Furthermore, cases may not excrete mucopolysaccharides for two to three months. Sometimes a clinical diagnosis cannot be made for two or three years. Not much can be done for the individual case, but diagnosis is desirable for use in genetic counseling. Therefore, the problem of diagnosis of mucopolysaccharidoses must be dealt with in the hospital or clinic laboratory on a case-by-case basis.

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* See Bulletins 53 and 56 on this subject.

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The Oregon program has included the Alpha-1 antitrypsin test to find potential cases of familial emphysema. However, there is difficulty in using this test productively in the newborn period so it has been discontinued. Here again is a procedure which may be important when it comes to genetic counseling but which is not productive in a routine screening program. A fruitful discussion appeared in the following: Sun, T., Kurtz, S., and Copeland, B. E., Alpha-1-antitrypsin Deficiency and Pulmonary Disease. Am. J. Clin. Pathology 62 : 725-731, Dec. 1974.

For Montana, an important benefit to be derived from the Oregon program is the availability of consultation. The persons who will usually be available are :

Rhesa L. Penn, Jr., M. D., Manager, Maternal and Child Health Section
Health Division, Department of Human Resources
1400 S. W. 5th Avenue, Portland, Oregon 97201
Telephone : 503 / 229-5593

Neil R. M. Buist, M. B., Ch. B.
Associate Professor of Pediatrics and Medical Genetics
Department of Pediatrics
University of Oregon Health Sciences Center
Portland, Oregon 97201
Telephones : 503 / 225-8392 and -8344
503 / 292-0237 (Home)

Gatlin R. Brandon, Director
Public Health Laboratory
Health Division, Department of Human Resources
1400 S. W. 5th Avenue, Portland, Oregon 97201
Telephone : 503 / 229-5466

The University of Oregon Hospital will accept patients from Montana under this program but, being from out of state, reimbursement will have to be arranged.

INSTRUCTIONS (other than those printed on the reverse of the filter paper)

1. Specimens are to be sent directly to Oregon : Oregon State Board of Health
Public Health Laboratory-1007
P. O. Box 231
Portland, Oregon 97207

2. Specimens are to be taken in conformity with the Oregon program which requires a specimen to be taken from each newborn before discharge from the hospital. If the patient has not been on milk for 48 hours when the specimen is taken, the desirability of returning for a repeat specimen not later than the 14th day of life should be emphasized.

3. Specimens must be mailed daily. We are experiencing some difficulty with a number of abnormal Beutler enzyme tests for galactose while normal levels are being obtained in the *E. coli* test. Some of this is probably due to delay in shipping the filter papers. The enzyme test depends on fresh specimens (less than 10 days old).

4. Great care should be exercised to prevent contamination of the filter papers. Some of the false positive reactions may be due to this. When I was in Portland, I saw some papers from Montana which had formula spilled on them. There was another instance in which a secretary had been hiding her sugared donuts in the drawer where supplies of specimen collecting kits were kept. Powdered formula in the air is also another possible source of contamination.

5. Significant findings will be reported by telephone from Oregon to the Chief, Maternal and Child Health Bureau, Montana State Department of Health and Environmental Sciences. He will then get in touch with the physician who submitted the specimen to report the case and for consultation.

6. A specimen with normal findings will be reported to the Laboratory Division of the Montana Department of Health and Environmental Sciences and we will report the result to the hospital on a roster. So in the normal course of events, no news will be good news.

7. As in times past, much of the field work in connection with infant screening will be handled by the Nursing Bureau of the Department. Questions regarding this aspect of the program may be addressed to Virginia Kenyon, Chief, Nursing Bureau - telephone 449-2076.

8. Oregon has been having another problem with specimens from Montana. Some have been coming in with the blood spots still wet. Please air dry the specimen at room temperature before mailing because wet spots increase the chance of bacterial growth during transit.

Dr. Phillip D. Pallister, Clinical Director, Boulder River School and Hospital has been in continual contact with the problem of inborn errors of metabolism in Montana. It is far from a "theoretical" one for this state. Here are some figures from his records :

34 patients with PKU who are residents in Boulder or are members of families who have patients in Boulder. Additionally, there are 8 or 9 other cases having no connection with Boulder.

2 cases of homocystinuria
8 cases of mucopolysaccharidoses
18 cases of congenital hypothyroidism

Other conditions resulting in mental retardation, etc. which could have been prevented :

14 cases of Rubella syndrome
15 cases of Kernicterus (Rh factor)

The number of live births in Montana is continuing to increase. For the first half of this fiscal year there were 6,201 live births compared to 5,654 and 5,453 for the same periods in F.Y. 1974 and 1973 respectively.

Our laboratory is engaged in another project which has a relationship to Mental Retardation. This is the screening of children for evidence of increased amounts of lead in their blood. For this fiscal year we have a grant from the Center for Disease Control to establish this capability in the state laboratory and to screen several hundred children. Dr. Guthrie has also developed a micro procedure for free erythrocyte porphyrins to use as a screening tool to detect evidence of lead poisoning. He found quite a few confirmed cases of lead poisoning in older institutions in New York State and in Ontario. Plans are to add this test to regional screening programs. Positives in the FEP screening test would then have a test for level of lead in their blood performed.

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